L Number	Hits	Search Text	DB	Time stamp
1	2	myelinopathy near5 periaxin	USPAT; US-PGPUB; DERWENT	2003/07/15 11:21
2	2	myelinopathy near9 periaxin	USPAT; US-PGPUB; DERWENT	2003/07/15 11:21
3	2	myelinopathy and periaxin	USPAT; US-PGPUB; DERWENT	2003/07/15 11:21
4	15	periaxin and mutation	USPAT; US-PGPUB; DERWENT	2003/07/15 11:23
5	15	periaxin	USPAT; US-PGPUB; DERWENT	2003/07/15 11:23
6	4	periaxin and neuropathy	USPAT; US-PGPUB; DERWENT	2003/07/15 11:25
7	7272	CMT or HNPP or DSS or CHN or RLS and periaxin	USPAT; US-PGPUB; DERWENT	2003/07/15 11:26
8	13711	(CMT or HNPP or DSS or CHN or RLS and periaxin) and mutation or polymorphism	USPAT; US-PGPUB; DERWENT	2003/07/15 11:26
9	472	(CMT or HNPP or DSS or CHN or RLS and periaxin) and ((CMT or HNPP or DSS or CHN or RLS and periaxin) and mutation or polymorphism)	USPAT; US-PGPUB; DERWENT	2003/07/15 11:26
10	315	((CMT or HNPP or DSS or CHN or RLS and periaxin) and ((CMT or HNPP or DSS or CHN or RLS and periaxin) and mutation or polymorphism)) and PCR	USPAT; US-PGPUB; DERWENT	2003/07/15 11:26
11	152	(((CMT or HNPP or DSS or CHN or RLS and periaxin) and ((CMT or HNPP or DSS or CHN or RLS and periaxin) and mutation or polymorphism)) and PCR) and alteration	USPAT; US-PGPUB; DERWENT	2003/07/15 11:27

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=> s myelinopathy (2a) CMT or HNPP or DSS or CHN or RLS
L1        8857 MYELINOPATHY (2A) CMT OR HNPP OR DSS OR CHN OR RLS

=> s l1 and periaxin
L2           0 L1 AND PERIAXIN

=> s l1 (2a) periaxin
L3           0 L1 (2A) PERIAXIN

=> s l1 and mutation
L4         506 L1 AND MUTATION

=> s l4 and periaxin
L5           0 L4 AND PERIAXIN

=> s l4 and PCR
L6          57 L4 AND PCR

=> s l6 and detect?
L7          37 L6 AND DETECT?

=> dup rem l7
PROCESSING COMPLETED FOR L7
L8           18 DUP REM L7 (19 DUPLICATES REMOVED)

=> d l18 ti so
L18 NOT FOUND
The L-number entered has not been defined in this session, or it
has been deleted.  To see the L-numbers currently defined in this
session, enter DISPLAY HISTORY at an arrow prompt (=>).

=> d l8 1-18, ti so

L8   ANSWER 1 OF 18  CAPLUS  COPYRIGHT 2003 ACS
TI   Molecular genetic characterization of the EWS/CHN and RBP56/
     CHN fusion genes in extraskeletal myxoid chondrosarcoma
```

SO Genes, Chromosomes & Cancer (2002), 35(4), 340-352
CODEN: GCCAES; ISSN: 1045-2257

L8 ANSWER 2 OF 18 MEDLINE
TI [Analysis of **mutations** in the chromosome 17p11.2 region in patients with Charcot-Marie-Tooth type 1 disease and in patients with tomaculous neuropathy].
Analiza mutacija u regionu hromozoma 17p11.2 kod osoba s oboljenjem Sarko-Mari-Tut, tip jedan, i osoba obolelih od tomakulozne neuropatije.
SO SRPSKI ARHIV ZA CELOKUPNO LEKARSTVO, (2002 Mar-Apr) 130 (3-4) 59-63.
Journal code: 0027440. ISSN: 0370-8179.

L8 ANSWER 3 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI Use of real-time **PCR** to **detect** heterozygote DNA duplication and deletion in genomic regions associated with disease, such as human chromosomes 17p11.2-12, 7q11.2, 15q11.2, 22q11 and Xq22
SO PCT Int. Appl., 23 pp.
CODEN: PIXXD2

L8 ANSWER 4 OF 18 EMBASE COPYRIGHT 2003 ELSEVIER SCI. B.V.DUPLICATE 1
TI CNS myelination and PLP gene dosage.
SO Pharmacogenomics, (2001) 2/3 (263-272).
Refs: 77
ISSN: 1462-2416 CODEN: PARMFL

L8 ANSWER 5 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 2
TI Sequence-specific **detection** of aristolochic acid-DNA adducts in the human p53 gene by terminal transferase-dependent **PCR**.
SO Carcinogenesis (Oxford), (January, 2001) Vol. 22, No. 1, pp. 133-140. print.
ISSN: 0143-3334.

L8 ANSWER 6 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI Rapid real-time fluorescent **PCR** gene dosage test for the diagnosis of DNA duplications and deletions
SO Clinical Chemistry (Washington, D. C.) (2000), 46(10), 1574-1582
CODEN: CLCHAU; ISSN: 0009-9147

L8 ANSWER 7 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 3
TI PMP22 Thr118Met is not a clinically relevant CMT1 marker.
SO Journal of Neurology, (September, 2000) Vol. 247, No. 9, pp. 696-700. print.
ISSN: 0340-5354.

L8 ANSWER 8 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI Real-time quantitative polymerase chain reaction. A new method that **detects** both the peripheral myelin protein 22 duplication in Charcot-Marie-Tooth type 1A disease and the peripheral myelin protein 22 deletion in hereditary neuropathy with liability to pressure palsies
SO Human Genetics (2000), 107(5), 494-498
CODEN: HUGEDQ; ISSN: 0340-6717

L8 ANSWER 9 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI Diagnosis of haploidy and triploidy based on measurement of gene copy number by real-time **PCR**
SO Human Mutation (2000), 16(5), 431-436
CODEN: HUMUE3; ISSN: 1059-7794

L8 ANSWER 10 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 4
TI Charcot-Marie-Tooth disease type 1A (CMT1A) and hereditary neuropathy with liability to pressure palsies (**HNPP**): Reliable **detection**

of the CMT1A duplication and **HNPP** deletion using 8 microsatellite markers in 2 multiplex **PCRs**.
SO International Journal of Molecular Medicine, (October, 2000) Vol. 6, No. 4, pp. 421-426. print.
ISSN: 1107-3756.

L8 ANSWER 11 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI Novel **PCR**-based diagnostic tools for Charcot-Marie-Tooth type 1A and hereditary neuropathy with liability to pressure palsies
SO Journal of the Peripheral Nervous System (1999), 4(2), 117-122
CODEN: JPNSFO; ISSN: 1085-9489

L8 ANSWER 12 OF 18 EMBASE COPYRIGHT 2003 ELSEVIER SCI. B.V.
TI Genotypic analysis of tumor suppresser genes PTEN/MMAC1 and p53 in head and neck squamous cell carcinomas.
SO Laryngoscope, (1998) 108/10 (1553-1556).
Refs: 17
ISSN: 0023-852X CODEN: LARYA8

L8 ANSWER 13 OF 18 MEDLINE
TI **PCR**-based strategy for the diagnosis of hereditary neuropathy with liability to pressure palsies and Charcot-Marie-Tooth disease type 1A.
SO NEUROLOGY, (1998 Mar) 50 (3) 760-3.
Journal code: 0401060. ISSN: 0028-3878.

L8 ANSWER 14 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 5
TI Determination of gene dosage at the PMP22 and androgen receptor loci by quantitative **PCR**.
SO Clinical Chemistry, (April, 1998) Vol. 44, No. 4, pp. 724-730.
ISSN: 0009-9147.

L8 ANSWER 15 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
TI An adhesion test system based on Schneider cells to determine genotype-phenotype correlations for mutated P0 proteins.
SO Genetic Analysis Biomolecular Engineering, (Oct., 1998) Vol. 14, No. 4, pp. 117-119.

L8 ANSWER 16 OF 18 CAPLUS COPYRIGHT 2003 ACS
TI PMP-22 gene duplications and deletions identified in archival, paraffin-embedded sural nerve biopsy specimens: correlation to structural changes
SO Acta Neuropathologica (1998), 96(1), 13-21
CODEN: ANPTAL; ISSN: 0001-6322

L8 ANSWER 17 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 6
TI Three novel **mutations** and a de novo deletion **mutation** of the DAX-1 gene in patients with X-linked adrenal hypoplasia congenita.
SO Journal of Clinical Endocrinology & Metabolism, (Nov., 1997) Vol. 82, No. 11, pp. 3835-3841.
ISSN: 0021-972X.

L8 ANSWER 18 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 7
TI Polymorphisms in the PMP-22 gene region (17p11.2-12) are crucial for simplified diagnosis of duplications/deletions.
SO Human Genetics, (1997) Vol. 99, No. 5, pp. 688-691.
ISSN: 0340-6717.

=> d l8 8, 10, 11, 13 ABS So

L8 ANSWER 8 OF 18 CAPLUS COPYRIGHT 2003 ACS
AB In Charcot-Marie-Tooth type 1A disease (CMT1A), heterozygosity for the peripheral myelin protein 22 (PMP22) duplication increases the gene dose from two to three, whereas, in hereditary neuropathy with liability to pressure palsies (**HNPP**), heterozygosity for the PMP22 deletion reduces the gene dose from two to one. Thirty-eight Norwegian patients with CMT1, 4 patients with **HNPP**, 15 asymptomatic family members, and 45 normal controls were studied using the ABI 7700 sequence **detection** system and the TaqMan method of real-time quant. polymerase chain reaction (**PCR**). Using a comparative threshold cycle (Ct) method and albumin as ref. gene, the gene copy no. by PMP22 gene duplication or deletion on chromosome 17p11.2-12 was quantified. The PMP22 duplication ratio ranged from 1.50 to 2.21, the PMP22 deletion ratio ranged from 0.44 to 0.55, and the PMP22 ratio in normals ranged from 0.82 to 1.27. All samples were run in triplicate, with a mean std. deviation of 0.07 (range 0.01-0.17). Thirty-four of thirty-eight CMT1 patients (89.6%) had the PMP22 duplication and the four **HNPP** patients had the PMP22 deletion. This was not found in any of the asymptomatic family members or the controls. Real-time quant. **PCR** is a sensitive, specific, and reproducible method for diagnosing PMP22 duplication and deletion. The method is fast, allowing 13 patients to be diagnosed in 2 h. It involves no radioisotopes and requires no post-**PCR** handling. In our opinion, real-time quant. **PCR** is the first method of choice in diagnosing PMP22 duplication and deletion.
SO Human Genetics (2000), 107(5), 494-498
CODEN: HUGEDQ; ISSN: 0340-6717

L8 ANSWER 10 OF 18 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 4
AB Charcot-Marie-Tooth disease (CMT) and hereditary neuropathy with liability to pressure palsies (**HNPP**) are the most frequent inherited disorders of the peripheral nervous system. They are clinically and genetically heterogeneous. A submicroscopic tandem duplication of 1.5 Mb in chromosome 17p11.2-12 comprising the PMP22 gene is found in 70.7% of autosomal dominant Charcot-Marie-Tooth type 1 (CMT1) patients. A reciprocal deletion is found in 87.6% of **HNPP** patients. The size of the typical CMT1A duplication is too small for classical cytogenetics and the whole region including the CMT1A-REP elements is sometimes too complex for a single DNA analysis method. We present results of a multiplex **PCR** of 8 microsatellite markers with multicolour fluorescence primer labelling followed by fragment analysis on an ABI 310 Prism analyzer to simplify the diagnostic procedure. Results for 24 patients can be obtained within 24 h. This method was applied on 92 DNA samples of unrelated patients carrying a typical CMT1A duplication previously confirmed by two colour fluorescence in situ hybridization (FISH, probe c132G8) and EcoRI/SacI Southern blotting (probe pLR7.8). Three alleles of three different sizes were clearly **detected** at least once in 88 of them (95.6%). Subsequently this analysis was applied on 312 Czech patients and revealed a CMT1A/**HNPP** rearrangement in 109 out of them.
SO International Journal of Molecular Medicine, (October, 2000) Vol. 6, No. 4, pp. 421-426. print.
ISSN: 1107-3756.

L8 ANSWER 11 OF 18 CAPLUS COPYRIGHT 2003 ACS
AB The majority of cases of Charcot-Marie-Tooth type 1A (CMT1A) and hereditary neuropathy with liability to pressure palsies (**HNPP**) are the result of DNA duplications and deletions resp. of a 1.5 Mb region on 17p11.2. The region contains the peripheral myelin protein 22 gene (PMP-22) and is flanked by homologous proximal and distal CMT1A-REP elements. The majority of duplications and deletions arise during meiotic recombination following misalignment and unequal crossing-over between the proximal and distal CMT1A-REP elements. The cross-over break-points are most frequently located within a 1.7 Kb hotspot of recombination and

produce novel duplication or deletion junctional CMT1A-REPs with unique restriction patterns. Here we describe the use of **PCR** based tests, which amplify a 3.6 Kb region including the 1.7 Kb hotspot from specific CMT1A-REPs, for the rapid diagnosis of CMT1A and **HNPP** patients. In an anal. of 96 CMT1A and 30 **HNPP** patients, duplication and deletion events were **detected** in all samples with cross-over breakpoints known to be within the region amplified by **PCR**.

SO Journal of the Peripheral Nervous System (1999), 4(2), 117-122
CODEN: JPNSFO; ISSN: 1085-9489

L8 ANSWER 13 OF 18 MEDLINE

AB Charcot-Marie-Tooth disease type 1A (CMT1A) and hereditary neuropathy with liability to pressure palsies (**HNPP**) are inherited peripheral neuropathies. In most cases these disorders are caused by either the duplication (in CMT1A) or the deletion (in **HNPP**) of a 1.5-megabase DNA fragment on chromosome 17p11.2, which contains the peripheral myelin protein 22 gene (PMP22). We developed a rapid and simple quantitative **PCR** assay for the **detection** of the CMT1A duplication or the **HNPP** deletion. The assay is based on the quantitative determination of the copy number of a 240-base pair DNA fragment from exon 4 of the PMP22 gene. Quantification was done on an automated fluorescence sequencer. Using this method we analyzed four families with the **HNPP** phenotype. In these families we identified the deletion in all affected individuals. To test the validity of the method, we compared the quantitative **PCR** results from 50 DNA samples, including 15 samples from individuals with **HNPP**, 15 samples from CMT1A patients, and 20 from normal controls, with the results obtained by Southern blot analysis. Concordant results were obtained in 49 of the 50 cases.

SO NEUROLOGY, (1998 Mar) 50 (3) 760-3.
Journal code: 0401060. ISSN: 0028-3878.

=> d his

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FILE 'BIOSIS, EMBASE, MEDLINE, LIFESCI, CAPLUS' ENTERED AT 10:55:13 ON 15 JUL 2003

L1 8857 S MYELINOPATHY (2A) CMT OR HNPP OR DSS OR CHN OR RLS
L2 0 S L1 AND PERIAXIN
L3 0 S L1 (2A) PERIAXIN
L4 506 S L1 AND MUTATION
L5 0 S L4 AND PERIAXIN
L6 57 S L4 AND PCR
L7 37 S L6 AND DETECT?
L8 18 DUP REM L7 (19 DUPLICATES REMOVED)

=> s myelinopathy and mutation
L9 44 MYELINOPATHY AND MUTATION

=> s myelinopathy
L10 379 MYELINOPATHY

=> s l10 and periaxin
L11 6 L10 AND PERIAXIN

=> d l11 1-6

L11 ANSWER 1 OF 6 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
AN 2001:114682 BIOSIS
DN PREV200100114682
TI **Periaxin** mutations cause recessive Dejerine-Sottas neuropathy.

AU Boerkoel, Cornelius F.; Takashima, Hiroshi; Stankiewicz, Pawel; Garcia, Carlos A.; Leber, Steven M.; Rhee-Morris, Laila; Lupski, James R. (1)
CS (1) Department of Molecular and Human Genetics, One Baylor Plaza, Room 609E, Houston, TX, 77030: jlupski@bcm.tmc.edu USA
SO American Journal of Human Genetics, (February, 2001) Vol. 68, No. 2, pp. 325-333. print.
ISSN: 0002-9297.
DT Article
LA English
SL English

L11 ANSWER 2 OF 6 EMBASE COPYRIGHT 2003 ELSEVIER SCI. B.V.
AN 2001063954 EMBASE
TI **Periaxin** mutations cause recessive Dejerine-Sottas neuropathy.
AU Boerkoel C.F.; Takashima H.; Stankiewicz P.; Garcia C.A.; Leber S.M.; Rhee-Morris L.; Lupski J.R.
CS Dr. J.R. Lupski, Dept. of Molec. and Human Genetics, One Baylor Plaza, Houston, TX 77030, United States. jlupski@bcm.tmc.edu
SO American Journal of Human Genetics, (2001) 68/2 (325-333).
Refs: 24
ISSN: 0002-9297 CODEN: AJHGAG
CY United States
DT Journal; Article
FS 008 Neurology and Neurosurgery
022 Human Genetics
LA English
SL English

L11 ANSWER 3 OF 6 MEDLINE
AN 2001143341 MEDLINE
DN 21090499 PubMed ID: 11133365
TI **Periaxin** mutations cause recessive Dejerine-Sottas neuropathy.
CM Erratum in: Am J Hum Genet 2001 Feb;68(2):557
AU Boerkoel C F; Takashima H; Stankiewicz P; Garcia C A; Leber S M; Rhee-Morris L; Lupski J R
CS Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX 77030, USA.
NC K08 DK02738 (NIDDK)
R01 NS27042 (NINDS)
SO AMERICAN JOURNAL OF HUMAN GENETICS, (2001 Feb) 68 (2) 325-33.
Journal code: 0370475. ISSN: 0002-9297.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
OS GENBANK-AF321191; GENBANK-AF321192; GENBANK-118200; GENBANK-145900; GENBANK-162500; GENBANK-605253; OMIM
EM 200103
ED Entered STN: 20010404
Last Updated on STN: 20030105
Entered Medline: 20010308

L11 ANSWER 4 OF 6 LIFESCI COPYRIGHT 2003 CSA
AN 2002:82782 LIFESCI
TI **Periaxin** Mutations Cause Recessive Dejerine-Sottas Neuropathy
AU Boerkoel, C.F.; Takashima, Hiroshi; Stankiewicz, P.; Garcia, C.A.; Leber, S.M.; Rhee-Morris, L.; Lupski, J.R.
CS Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA
SO American Journal of Human Genetics [Am. J. Hum. Genet.], (20010200) vol. 68, no. 2, pp. 325-333.
ISSN: 0002-9297.
DT Journal
FS N3

LA English
SL English

L11 ANSWER 5 OF 6 CAPLUS COPYRIGHT 2003 ACS
AN 2002:504902 CAPLUS
DN 137:77308
TI Defects in **periaxin** associated with **myelinopathies** leads to diagnostic and drug screening applications
IN Lupski, James R.; Boerkoel, Cornelius F.; Takashima, Hiroshi
PA Baylor College of Medecine, USA
SO PCT Int. Appl., 99 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2002051981	A2	20020704	WO 2001-US48935	20011213
	W: CA, JP				
	US 2003039987	A1	20030227	US 2001-21955	20011213
PRAI	US 2000-255217P	P	20001213		

L11 ANSWER 6 OF 6 CAPLUS COPYRIGHT 2003 ACS
AN 2001:139619 CAPLUS
DN 135:120627
TI **Periaxin** mutations cause recessive Dejerine-Sottas neuropathy
AU Boerkoel, Cornelius F.; Takashima, Hiroshi; Stankiewicz, Pawel; Garcia, Carlos A.; Leber, Steven M.; Rhee-Morris, Laila; Lupski, James R.
CS Departments of Molecular, University of California Davis Health System, Sacramento, CA, USA
SO American Journal of Human Genetics (2001), 68(2), 325-333
CODEN: AJHGAG; ISSN: 0002-9297
PB University of Chicago Press
DT Journal
LA English
RE.CNT 24 THERE ARE 24 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

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     FILE 'MEDLINE, CAPLUS' ENTERED AT 15:36:30 ON 15 JUL 2003
L1            42 S PERIAXIN
L2            41 S L1 AND MYELIN?
L3             1 S L2 AND MYELINOPATHY
L4            20 S L2 AND (MUTATION? OR POLYMORPHISM? OR SUBSTITUTION? OR DELETI
L5            13 DUP REM L4 (7 DUPLICATES REMOVED)
L6            20 S L-PERIAXIN OR S-PERIAXIN
L7             0 S L6 NOT L1
L8             4 S PERIAXIN AND MAG
L9             4 DUP REM L8 (0 DUPLICATES REMOVED)

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